DISEASE:
Autosomal dominant limb-girdle muscular dystrophy type 1C

NAME:	Autosomal dominant limb-girdle muscular dystrophy type 1C
DESCRIPTION:	A rare subtype of autosomal dominant limb girdle muscular dystrophy characterized by a childhood to adulthood onset of progressive, mild-to-moderate proximal muscle weakness, calf hypertrophy, and variable muscle cramping/stiffness or myalgia, after exercise. A positive Gowers sign and elevated creatine kinase serum levels are frequently observed. Initial motor milestones are usually normal and muscle rippling may be observed. Respiratory and cardiac anomalies are generally not associated.
ORPHACODE:	265
SYNONYMS:	LGMD1C Limb-girdle muscular dystrophy due to caveolin-3 deficiency
XREF(S):	Orphanet OMIM ICD-10 OMIM
ANALYTE(S):	CAV3
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